

Original Research Article

Clinical Profile of Congenital Structural Defects in Pregnant Women Seeking for Medical Termination of Pregnancy in Second Trimester of Pregnancy at A Teaching Hospital.

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Abstract:

Introduction: Any irreparable aberration in the physical, anatomical, or functional form present in an unborn child before birth is referred to as a congenital malformation. Advancement in ultrasonography leads to more and more detection of these anomalies. Due to improved imaging capabilities, prenatal malformations are now discovered earlier, giving antenatal care providers or doctors the chance to provide counselling with the option of terminating the pregnancy in situations of lethal defects.

Material and methods: This is a prospective observational study done at tertiary care teaching hospital. Period of the study was one year from January to December 2022. During this period total 513 cases came for medical termination of pregnancy at tertiary care centre out of which 131 cases were enrolled for the study as per the inclusion criteria. All those pregnancies were included in study population who were between 13-to-28-week gestational weeks, and came with ultrasonography report of congenital structural defect in fetus. Epidemiological data like age, residence, their gravida, type of structural defects, history of previous cesarean sections was noted and analysed for their distribution and presentation.

Results: in our study it was found that most of the cases were primigravida, of age 20 -30 years, and having most common anomalies belong to central nervous system followed by multiple organ system involvement. Most of CNS anomalies were anencephaly, hydrocephalus, and acrania exencephaly sequence.

Conclusions: Birth defects rank among the most frequent grounds given by couples choosing second trimester. In our study, the combination of anencephaly and acrania anencephaly was one of the main factors of birth defects reported to a centre that could be quickly identified by experienced sonologists in the late first trimester and could be terminated within safe limits, further keeping antenatal women within a safe window of medical termination of pregnancy and lowering maternal morbidity.

Keywords:

Congenital structural defect, Anencephaly, acrania, medical termination of pregnancy.

Introduction:

Any irreparable aberration in the physical, anatomical, or functional form present in an unborn child before birth is referred to as a congenital malformation.¹ Every year, an estimated 2,40,000 new-borns die within the first 28 days of life owing to congenital diseases. Congenital diseases kill an additional 170,000 children between the ages of one month and five years.²

Advancement in ultrasonography leads to more and more detection of these anomalies. Visualising embryonic and early foetal anatomy has been significantly enhanced by significant advancements in signal processing and imaging magnification.³

Due to improved imaging capabilities, prenatal malformations are now discovered earlier, giving antenatal care providers or doctors the chance to provide counselling with the option of terminating the pregnancy in situations of lethal defects.³

Many women are receiving a foetal anomaly diagnosis quite late, often in the second trimester of pregnancy, as a result of lack of information about prenatal screening and diagnostic testing.⁴ Three categories of congenital foetal malformations—lethal, severe, and mild—can be distinguished. Major congenital abnormalities are those that are fatal or severe, whereas moderate flaws are considered minor defects.⁵

According to the Medical Termination of Pregnancy Act of 1971 and its amendment, all those pregnancies that have a substantial risk assessment that, if the child were to be born, it would have severe physical or mental abnormalities are permitted to be terminated by the registered medical practitioner.⁶ As per the new amendment pregnancy from 21 to 24 week can be terminated under various clauses among it one of, the clause state that a pregnancy with substantial risk of life to fetus or major handicap in future is allowed to terminate.

After the amendment there was a sudden surge of patient for medical termination of pregnancy in the present hospital so the study designed to study pattern of congenital malformation in both gestational age group defined as in MTP act 2021 amendment i.e., 13 to 20 weeks and 21 to 24 weeks. so the study was designed to know the distribution of cases, organ system involved and clinical demographics of cases who opted for second trimester pregnancy termination due to structural birth defects.

Material and method:

This is a prospective observational study done at tertiary care teaching hospital. Period of the study was one year from January to December 2022 at Pandit Bhagwat Dayal Sharma Postgraduate institute of medical sciences, Rohtak, Haryana. During this period total 513 cases came for medical termination of pregnancy at tertiary care centre out of which 131 cases were enrolled for the study as per the inclusion criteria.

Inclusion criteria:

1. Pregnancy should be of second trimester (13-28 weeks)
2. Pregnancy should have major congenital birth defect diagnosed by institutional ultrasonography

Exclusion criteria:

Second trimester MTP without structural defects, Epidemiological data like age, residence, their gravida, type of structural defects, history of previous cesarean sections was noted and analysed for their distribution and presentation.

Results:

In our study majority of cases belong to 20-to-25-year age group, and most of them of rural background, approximately all cases from state Haryana except 3 case which were outside of Haryana as shown in table 1.

As per table 2 most of the cases were primigravida i.e., 37.43%, on observing the distribution of cases majority of cases were of 13 to 20 weeks which closely follows the gestational age 21-to-24-week gestational age i.e., 55.38 and 41.98 respectively.

Among all cases 10 cases were with history of prior cesarean section of which one case with history of LSCS.

As per table 3 maximum number of cases were with the involvement of fetal central nervous system (CNS) which followed by multiple system involvement i.e., 14.50%.

Most of CNS anomalies were anencephaly, hydrocephalus, and acrania exencephaly sequence. In most cases, multiple organ involvement was associated with CNS and cardiovascular system involvement, or numerous abnormalities in combination with musculoskeletal system involvement as shown in table 4.

Table 1: Epidemiological Profile

Age (years)		
15-20	16	12.21
21-25	60	45.80
26-30	33	25.19
31-35	18	13.74
>35	4	3.053
Residence		
Urban	45	34.3
Rural	86	65.6
Haryana	128	97.7
Other states	3	2.29

Table 2: Obstetric Profile:

gravida	n	%
G1	49	37.40
G2	34	25.95
G3	28	21.37
G4	9	6.87
>G4	11	8.39
Gestational age		
13+6 -20w+6D	66	55.38
21-24w	55	41.98
24w+1d -28w	10	7.63
History of cesarean section		
Yes	10	7.63
no	121	92.3
Order of LSCS		
Previous one	7	5.34
Previous two	2	1.52
Previous three	1	0.76

Table 3: Distribution of Structural birth defect as per the system involved:

CNS	82	62.59
CVS	9	6.87
GUT	5	3.81
GIT	6	4.58
Musculoskeletal	10	7.63
Others	19	14.50

Table 4: Anomalies Distribution

CNS	Total cases (n)	%
Anencephaly	26	19.84
hydrocephalus	13	9.92
Iniencephaly	1	0.76
Exencephaly	3	2.29
Encephalocele	8	6.10
Acrania exencephaly sequence	12	9.16
Meningomyelocele	8	6.10
Arnold Chiari Malformation	5	3.81
Holoprosencephaly	1	0.76
Spinal dysraphism	2	1.52
Dandy walker phenomenon	3	2.29
CVS		
Hypoplastic left ventricle	4	3.05
TGA	1	0.76
Tetralogy of Fallot	2	1.52
VSD	1	0.76
Truncus Arteriosus	1	0.76
Renal		
MCKD	3	2.29
Renal agenesis	1	0.76
Megacystis	1	0.76
Abdomen		
Omphalocele	1	0.76
Anterior abdominal wall defect	2	1.52
Congenital diaphragmatic hernia	3	2.29
Musculoskeletal system		
Short limbs	3	2.29
Skeletal dysplasia	6	4.58
Osteogenesis imperfecta	1	0.76
Multiple organ involvement	1	0.76
Hydrops fetalis	3	2.29
Conjoint twins	1	0.76

Discussion:

As per the Medical termination of pregnancy act 1971 abortions are allowed in India. Any pregnancy can be terminated if the act's provisions are followed. Substantial risk to life to the fetus is one of the major reasons for couples to opt for termination of pregnancy in Haryana region most of the second trimester pregnancy termination is being done under the clause mention in act "There is a significant risk that the child, if born, would have such physical or mental abnormalities as to be severely

handicapped.” Substantial fetal anomalies are those anomalies which carries poor prognosis either intrauterine or extrauterine on continuation of pregnancy.⁷

In our study the cases were of mainly 21 to 25 years age while study done at Brazil for second trimester of pregnancy with structural defect also observed the age group of 19 to 29 years. Which suggests most of the females were young and might have less knowledge regarding available sonography to detect structural defects at earlier gestational age group.⁸ On observing their residence it has been seen that 65.6 % cases belong to rural area which contrasts with study done by Kakkar A et al. in year 2021 where most of the cases were of urban area although that study was done only in national capital region of India which could be the reason that their cases were urban while present study was done at second tier city Rohtak which shows the difference in population.⁹ In present study most of the cases were of primigravida while study done by Gupta S et al which was a cross sectional study done in city Jamnagar State Gujrat patient were mostly gravida order two which shows the difference in presentation of cases.⁹ The current study only included MTP cases that were in the second trimester and had congenital anomalies, which may have contributed to the differences in parity presentations. Study done by Kakkar A et al. also shows that their cases were mainly gravida 2 instead of being primigravida in our study which indirectly shows concern that primigravida cases are not participating in good antenatal care despite of free antenatal care provisions by government of India.¹⁰

Present study includes 55.38% cases in early 2nd trimester while 41.98% cases belong to 21 to 24 weeks as there were very scarce studies done in India after the amendment of MTP act 2021 out of all 131 second trimester cases 41.98% cases were of 21 to 24 weeks pregnancy while 7.6% cases were beyond 24 weeks. All the termination done for substantial risk of fetal abnormalities. It was our departmental protocol to take pediatric surgeon opinion before deciding the condition is with substantial risk is there or not while in certain cardiovascular abnormalities opinion of pediatric cardiologist was also take before proceeding towards medical termination of pregnancy. Decisions were made under medical board for pregnancy beyond 24 weeks as determined by ultrasound and dated by last menstrual cycle and reported with foetal structural defects. Its members include a gynaecologist, a paediatrician, and a radiologist.^{6,7}

If the board finds that the woman is fit for termination of pregnancy and that ultrasonography proved foetal structural abnormalities is carrying significant risk to terminate the pregnancy, then only termination of pregnancy was conducted. only 10 medical board were carried out during the study period and in those cases, anomalies were of central nervous system mainly anencephaly. As per research done on PubMed and Google this is the first stud which was conducted to know the profile of cases who were seeking for MTP beyond 20 weeks along with second trimester clinical case profile. On examining the profile of the case most structural anomalies which were detected during the second trimester were of central nervous system (62.59%) which followed by multiple structural abnormalities which include more than one organ system i.e., 14.50% which is in like study done by Rydberg C, Tunón K. where they also found highest structural anomaly in fetal CNS.¹¹ On examining the CNS anomalies most of the CNS anomalies were anencephaly and acrania exencephaly sequence besides that it was also observed that hydrocephalus was another condition which is catering quite high number among rest of the structural abnormalities. Which Indicates that patients are reporting very late to radiologists. Amongst ese anencephaly can be easily diagnosed in late first trimester which is supported by the evidences given by Porath M et al. who suggested that at 12 to 13 weeks gestation, anencephaly is appropriately detected. Anencephaly occurs when the cranial vault is absent. Until the initiation of ossification has been definitively failed, ultrasound results might be normal. At 12 to 13 weeks, a first trimester scan allows for accurate diagnosis and aggressive care of anencephaly.¹²

Among CVS Anomalies hypoplastic left heart syndrome was seen in 4 cases which is one of the serious conditions and life-threatening substantial anomaly where one of the options offered to the couple is MTP as suggested evidence study done by Claxon-McKinney B.¹³

Besides that, 7.63% cases were diagnosed with musculoskeletal abnormalities among musculoskeletal structural abnormalities 6(4.58%) cases were of skeletal dysplasia. Lethal skeletal dysplasia are the genetic conditions where abnormal growth and development of bone and cartilage occurs. Although prompt termination of pregnancy was advised to the couples with simultaneous counselling for further genetic testing and autopsy of fetus, all the couples denied for autopsy due to cultural reasons.¹⁴

Conclusion:

Birth defects rank among the most frequent grounds given by couples choosing second trimester pregnancy. Contrary to first trimester abortion, second trimester abortions are more difficult, it becomes riskier due to the foetus' close relationship with the mother's tissues. Despite efforts done by the government for good antenatal care still there is delay in reaching to health care facilities which further delays in prenatal diagnosis of many congenital malformations which can be diagnosed in first trimester easily.

In our study, the combination of anencephaly and acrania anencephaly was one of the main factors of birth defects reported to a centre that could be quickly identified by experienced sonologists in the late first trimester and could be terminated within safe limits, further keeping antenatal women within a safe window of medical termination of pregnancy and lowering maternal morbidity.

Conflicts of interest: none

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